



App No.: 09/693,205
Title: Identification of ARSACS ...
Inventors: Thomas J. Hudson, *et al.*

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O.G. FIG.			
APPROVED	BY	CLASS	SUBCLASS
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Chromosome 13

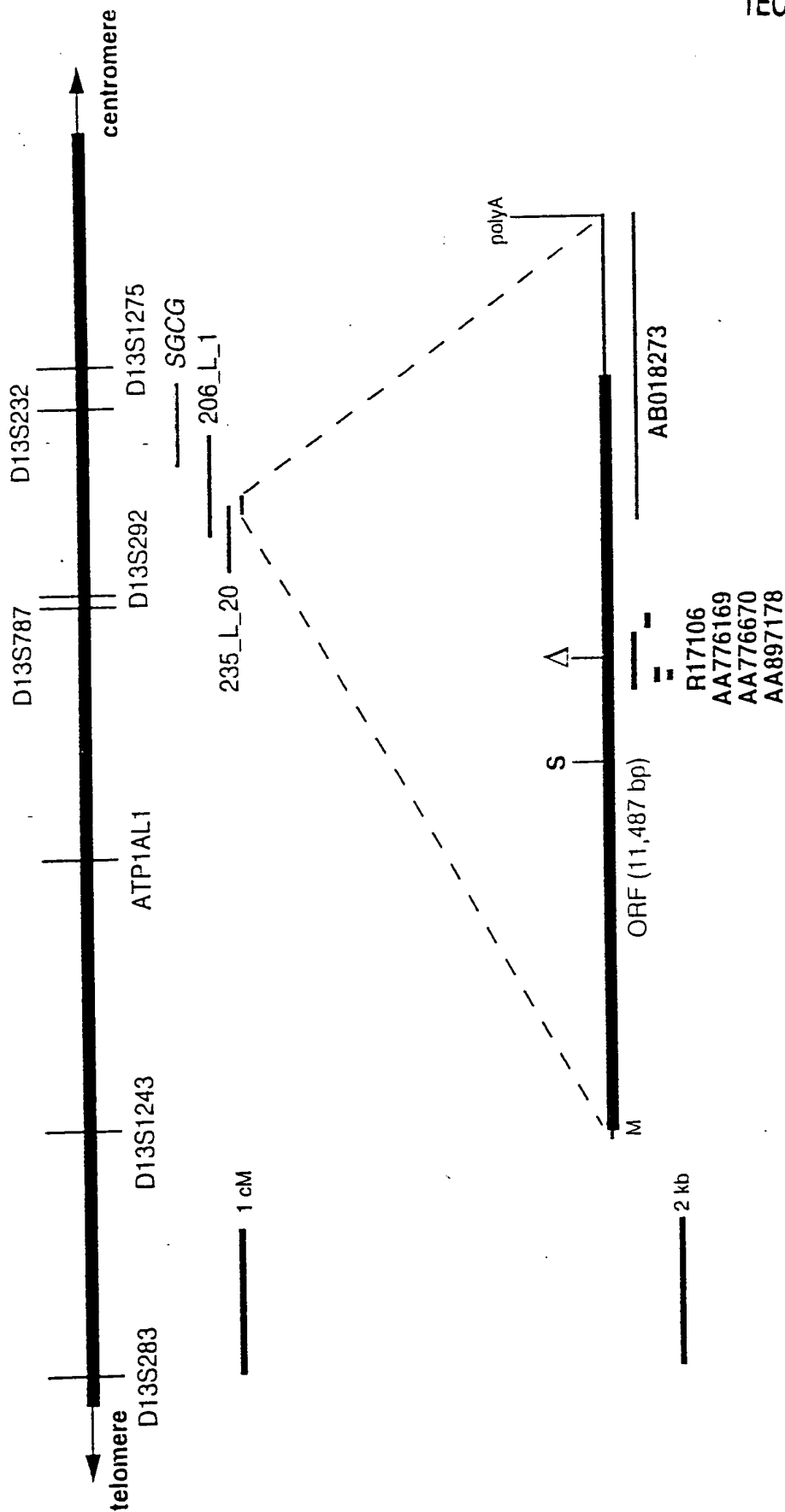


Figure 1



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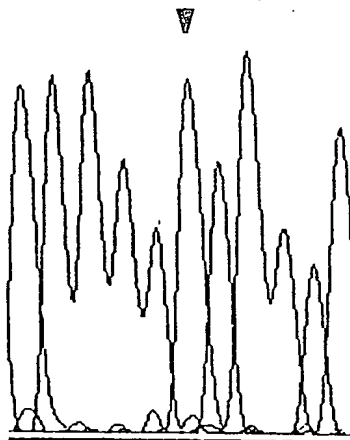
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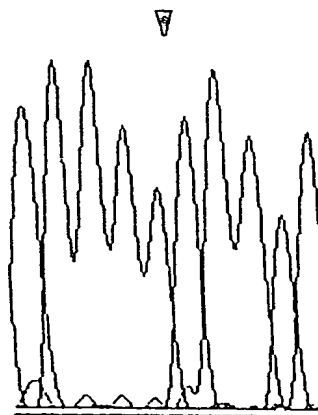
APPROVED BY DRAFTSMAN	O.G. FIG.	SUBCLASS
	CLASS	

ACCCTATTCA
(SEQ ID NO: 17)



6594T

ACCCTATTCA
(SEQ ID NO: 18)



6594Δ

Figure 2A



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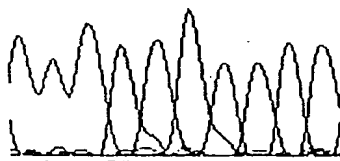
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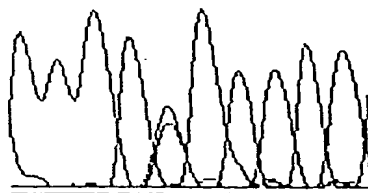
APPROVED BY DRAFTSMAN	CLASS	SUBCLASS
	O.G. FIG.	

A A A G C G A C A C
(SEQ ID NO: 19)



5254C

A A A G N G A C A C
(SEQ ID NO: 20)



5254C/T

Figure 2B



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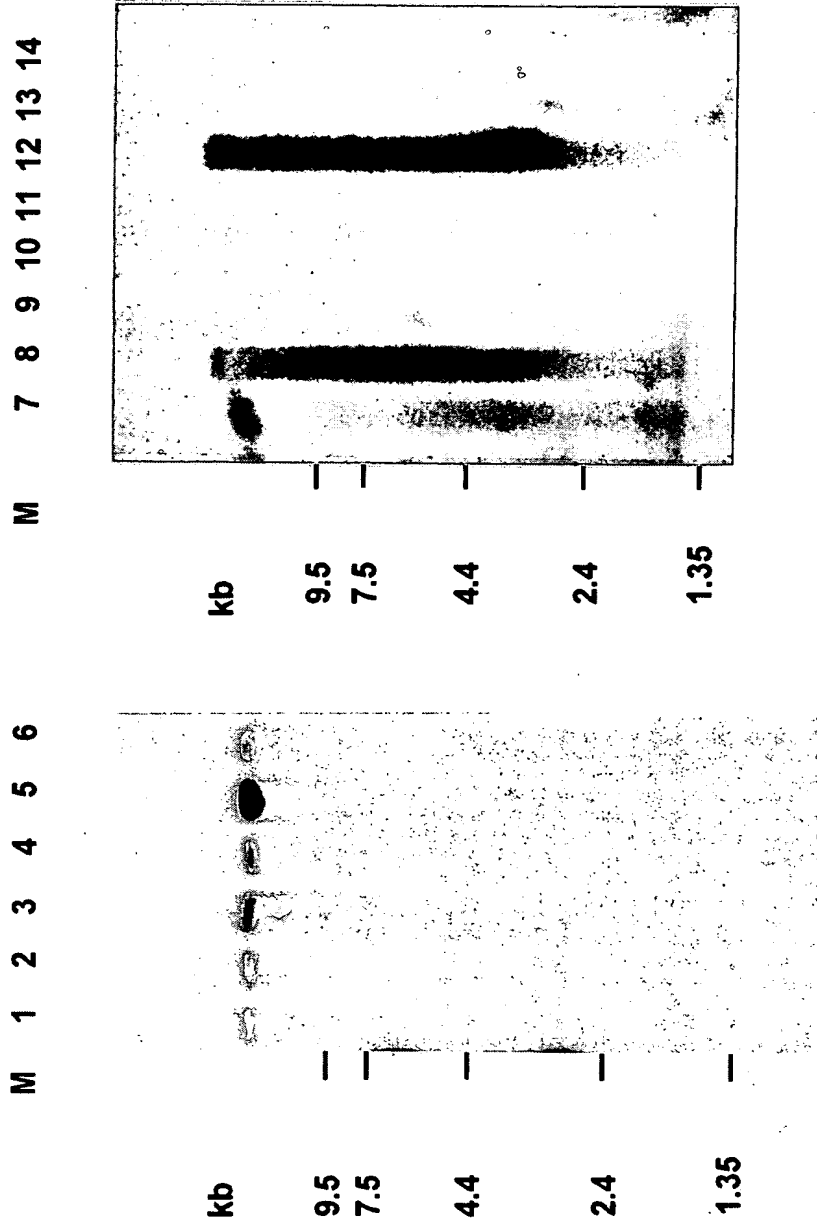


Figure 3

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APPROVED		O.G. FIG.	
BY	CLASS	BY	SUBCLASS
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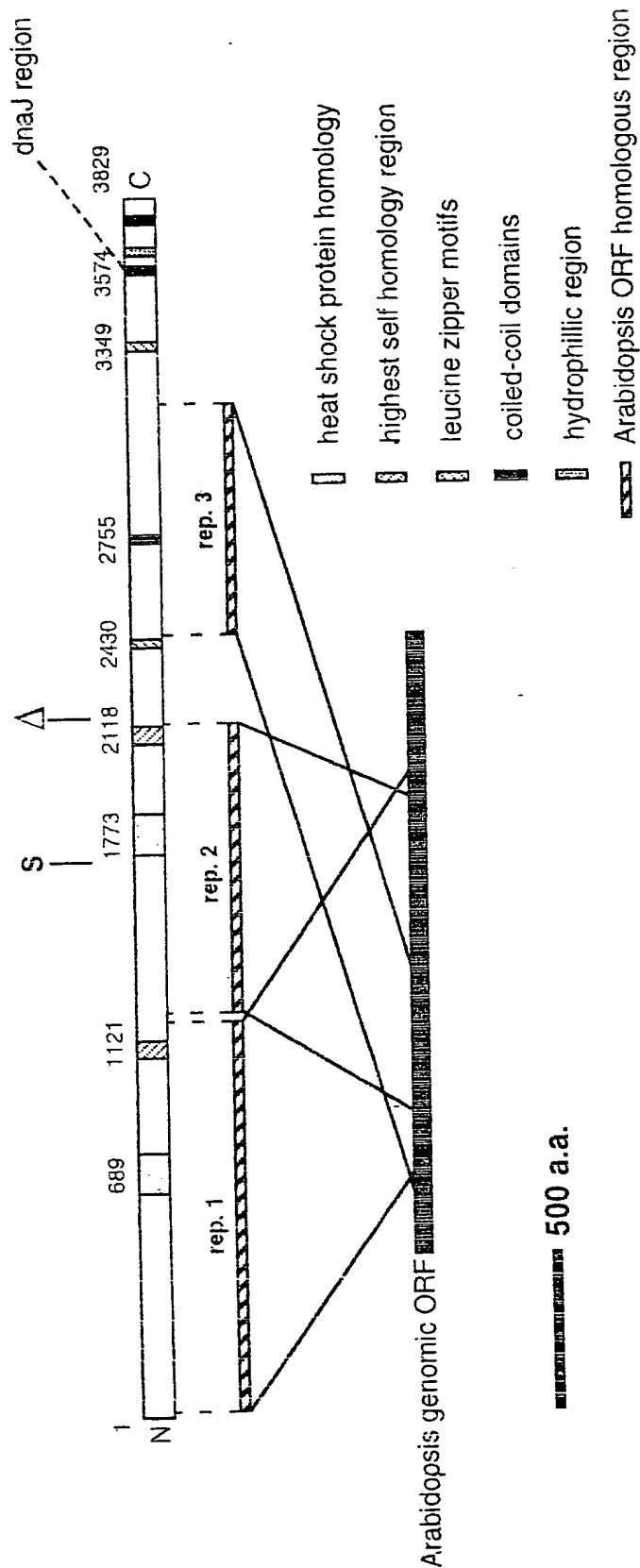


Figure 4A



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APPROVED	O.G. FIG.	
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HUMAN-REP 1	705	GOREPLTVRIKNILEEYPSVSDIFKEILQONADOAN-ATECSFLIDMRNMDIRENLLDPG
MOUSE-REP 1	705	GOREPLTVRIKNILEEYPSVSDIFKEILQONADOAN-ATECSFLIDMRNMDIRENLLDPG
HUMAN-REP 2	1773	Gqke.LT.RIK.IL.YPS.em.KEILQONADOA..ATE..Fv.D.R.....d.i.D..
ARABIDOPSIS	987	Gqhe.LT.RIK.ILE.Y.....I..EIYQNAEOA..ASE.tFLID.h.....d.LL.P.
HSP90 (YEAST)	18	.Q.E..l....I...Y.....rELi.NA.OA.....L.D.k.....d.i....
HUMAN-REP 1	764	MAACHGPALWSFNNSQ-FSDSDFVN----ITRLG-----ESLKRGE-VDKVGKEGLGFNS
HUMAN-REP 1	764	MAACHGPALWSFNNSQ-FSDSDFIN...ITRLG.....ESLKRGE-VDKVGKEGLGFNS
HUMAN-REP 2	1830	A...GPAL..yNN...Fe.D.....I.LG.....K.G....K.G.YGIGFNS
ARABIDOPSIS	1044	MA...GPALy.FNNS.FE.D.....ISriG.....qa.K.....igrPGLGFN.
HSP90 (YEAST)	78L...q.....L.ae.in....I.k.G.....EaL..G..V..iG.FGYGF.S
HUMAN-REP 1	813	VYHITDIPiIMSR-----EFMIMFDP
MOUSE-REP 1	813	VYHITDIPiIMSR.....EFMIMFDP
MOUSE-REP 2	1879	VYHITD.P..iS.....d.l.iFD.
ARABIDOPSIS	1093	VYH.TDIP..vS.....E.ivMFDP
HSP90 (YEAST)	137	If.v.D...visk.....F.v..D.

Underline = conserved
Bold = identical

Figure 4B



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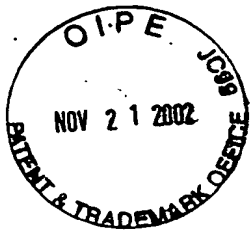
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APPROVED	O.G. FIG.	
	CLASS	SUBCLASS
BY	DRAFTSMAN	

MNTFWPGRELIVQWYPFDENRNHPSVSWLKMVWKNLYIHFSEDLTLFDEMPLIPRTILEEV.....S.DKR...L.....L.N.	60
GQTCVELIRLRIPSLVILDDESEALPEFLADIVQKLGGFVLKLDASIQHPLIKKYIHS D.....V.....T.....I...R...T.....V.....	120
PLPSAVLQIMEKMPLQKLCNQITSLLPTHKDALRKFLASLTDSSSEKEKRIIQELAIFKRII.....I.....A.....T.....T.....	180
NHSSDQGISSYTKLKGCKVLHHTAKLPADLRLSISVIDSSDEATIRLANMLKIEQLKTTD.....T.....V.....K.....	240
CLKLVLKDIENAFYSHEEVTLQMLWVLENLSSLKNNPNVLEWLTPLKFIQISQEQMVSA ...F....G....TQ.....I.....S...D..M....HM..GHV.A.	300
GELFDPDIEVLKDLFCNEEGTYFPSPVFTSPDILHSLRQIGLKNEASLKEKDVVQVAKKI .D.....R...Y...EAC...TI.....S.....R..	360
EALQVGACPDQDVLKKAKTLLLVLNKNHTLLQSSEGMKTLKKIKWVPACKERPPNYPGSSS.QN....M.....Q.....A.....	420
LVWKGDLNLCAPPDMCDVGHAILIGSSSLPLVESIHVNLEKALGIFTKPSLSAVLKHFKIAA..V.V.....V....Q..S.....TIN.....T	480
VVDWYSSKTFSDYQFHILLEIYGFMDHLNEGKDSFRALKFPWVWTGKKFCPLAQT.....S.....K.....N.....	540
VIKPIHDLDLQPYLHNVPKTMKFHQLFKVCGSIEELTSDHISMVIQKIYKSDQDLSEQ ...T.....Y.....A.....V.....E...E	600
ESKQNLHMLNIIIRWLYSNQIPASPNTVPPIHHSKNPSKLIMKPIHECCYCDIKVDDLNDM.....Y..R....V.....	660
LLEDSEVEPIILVHEDI PMKTAEWLKVPCSLSTRLINPENMGFEQSGQREPLTVRIKNILEE	720
YPSVSDIFKELLQNADDANATECSFLIDMRRNMDIRENLLDPGMAACHGPALWSFNNSQFM.....E.	780
SDSDFVNITRLGESLKRGEVDKVGKFGGLGFNSVYHITDIPIMMSREFMIMFDPNINHISKL.....	840
HIKDKSNPGIKINWSKQKRLRKFPNQFKPFIDVFGCQLPLTVEAPYSYNGTLFRLSFRT ...R.....A.....	900
QQEAKVSEVSSTCYNTADIYSLVDEFSLCGHRLIIFTQSVKSMYLYKIEETNP SLAQDN.....	960
TVIIKKKSCSSKALNTPVLSVLKEAAKLMKTCSSSNKKLPSDEPKSSCILQITVEEFHHV .I....V.P....A.....T.V.....	1020
FRRIADLQSPLFRGPDDPAALFEMAKSGQSKKPSDELSQKTVECTTWLLCTCMDTGEALT.....P....D....I.....	1080
KFSLSESGRRLGLVPCGAVGVQLSEIQDKWTVKPHIGEVFCYLPRIKTGLPVHINGCF ...N.....L.H.T.E.....I.....	1140
AVTSNRKEIWKTDTKGRWNTTFMRHVIVKAYLQVLSVLRDLATSGELMDYTTYAVWDPDPA.....IG...T.....	1200
LVHDDFSVICQGFYEDIAHGKGKELTKVFSDBGSTWVSMKNVRLDDSIKRRDVGSAAFKK.....R.....M.....Q.K.....	1260
IFLKYLKKTGSKNLCAVELPSSVKLGFEAGCKQILLENTFSEKQFFSEVFFPNIQEIEAA.....	1320
ELRDPLMIFVLNEKVDEFSGVLRVTPCIPCSLEGHPLVLP SRLIHPEGRVAKLFDIKDGRN.....L....I.....V.....T.....	1380
FPYGSTQDYLNPIILI KLVLQLGAKDDILWDDMLERAVSVAEINKSDHVAACLRSSILLSM.....E.....A.....	1440

Figure 5A



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APPROVED BY DRAFTSMAN	O.G. FIG.	
	CLASS	SUBCLASS

LIDEKLRKIRDPRAKDFAAKYQTIRFLPFLTKPAGFSLDWKGNFSPKPMFAATDLYTAEHK.....P.....E.....I.....Y	1500
QDIVCLLQPILNENSHSFRGCGSVSLAVKEFLGLLKPTVDLVINQLKEVAKSVDDGITLQ.....	1560
YQENITNACYKYLHEALMQNEITKMSIIDKLPFSFILVENAYVDSEKVSFHLNFEEAAPYVL...MA.AT..E.....C.....V..E.....	1620
LYQLPNKYKNNFRELFTVGVRSCTVEDFALVLESIDQERGKQITEENFQLCRRIISES.....F.....K.....	1680
GIWSLIREKKQEFCEKNYKILLPDTNLMLLPAKSLCYNDPCWIKVKDTTVKYCHADIPRR.....L.....S.....	1740
EVAVKLGAVPKRHKALERYASNVCFTTLGTEFGQKEKLTSRIKSILNAYPSEKEMLKELLI.....I...A.....	1800
QNADDAKATEICFVFDPRQHPVDRIFFDDKWAPLQGPALCVYNNQPFTEDDVRGIQNLGKG	1860
TKEGNPYKTGQYGIGFNSVYHITDCPSFISGNDILCIFDPHARYAPGATSISPGRMFRDLC...H.....G.....V.....	1920
DADFRTQFSDVLDLYLGTHFKLDNCTMFRFPLRNAEMAKVSEISSVPASDRMVQNLLDKLN.....Q.....S.....	1980
RSDGAELLMFLNHMEKISICEIDKSTGALNVLYSVKGKITDGDRLKRKQFHASVIDSVTKA..G.....	2040
KRQLKDIPVQQITYTMDTEDSEGNLTWILICNRSGFSSMEKVSKSVISAHKNQDITLFPR	2100
GGVAACITHNYKKPHRAFCFLPLSLETGLPFHVNGHFALDSARRNLWRDDNGVGVRSDWN	2160
NSLMTALIAPAYVELLIQLKKRYFPGSDPTLSVLQNTPIHVVKDTLKKFLSFFPVNRLDL	2220
QPDLYCLVKALYNCIHEDMKRLLPVVRAPNIDGSDLHSAVITWINMSTSNKTRPFFDNLS.....	2280
LQDELQHLKNADYNITTRKTVAENVYRLKHLLEIGFNLVYNCDETANLYHCLIDADIPVV.....	2340
SYVTPADIRSFLMTFSSPDNTCHIGKLPCLQQTNLKLHSLKLLVDYCFKDAEENEIEVV.....S.F..	2400
EGLPLLITLDSVLQTFDAKRPKFLTTYHELIPSRKDLFMNTLYLKYSNILLNCKVAKVFDI..G.....SV.....	2460
ISSFADLLSSVLPREYKTKSCTKWKDNFASWSLKNAWHFISESVSVKEDQEETKPTFDIN.A.....TD...P..A..V	2520
VVDTLKDWALLPGTKFTVSANQLVVPEGDVLLPLSLMHIAVFPNAQSDKVFHALMKAGCI I..I.....TS.....I.....	2580
QLALNKICSKDSAFVPLLSCHTANIESPTSILKALHYMVQTSTFRAEKLVENDFEALLMYL.....D..A.....V.....T...M.....	2640
FNCNLNHLMSQDDIKILKSLPCYKSISGRYVSIGKFGTCYVLTKSIPSAEVEKWTQSSSSS.....M..A.....	2700
AFLEEKIHLKELYEVIGCVPVDDLEVYLKHLPLKIENTLSYDAKLEHLIYLNRLSSAEELV.....L.....A.I..P	2760
<u>SEIKEQLFEKLESLLIIHDANSRLKQAKHFYDRTVRVFEVMLPEKLFIPNDFFKLEQLI</u>N.....KE.....V.	2820
KPKNHVTFMTSWVEFLRNIGLKYILSQQLLQFAKEISVRANTENWSKETLQNTVDILLHQAA.....A.....S.....	2880

Figure 5B



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APPROVED BY DRAFTSMAN	O.G. FIG.	SUBCLASS
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HIFQERMDLLSGNFLKELSLIPFLCERAPAEFIRFHPQYQEVNGTLPLIKFNGAQVNP	2940
.....Y.....	
FKQCDVLQLLWTSCPIPEKATPLSIEQEGSDLGPPQEQLQVLNMLNVNLDPPDKVIN	3000
.....A.....	
NCRNICNITTLDEEMVKTRAKVLRSIYEFLSAEKREFRFQLRGVAFVMVEDGWKLLKPEE	3060
.....	
VVINLEYESDFKPYLYKLPLELGTTFHQLFKHLGTEDIISTKQYVEVLSRIFKNSEKQLD	3120
.....A.....S.....	
PNEMRTVKRVVSGFLRSLQNDSVKVRSDLENVRDLALYLP SQDGRLVKSSILVFDDAPHY	3180
.....K.....A.....K.....	
KSRIQGNIGVQMLVDLSQCYLGKDHGFHTKLIMLFPQKLRPRLLSSILEEQLEETPKVC	3240
.....	
QFGALCSLQGRQLQLLSSEQFITGLIRIMKHENDNAFLANEEKAIRLCKALREGLKVSCF	3300
.....	
EKLQTTLRVKGFNPPIPHSRSETFAFLKRFGNVILLYIQHSDSKDINFLALAMTLKSAT	3360
.....	
DNLISDTSYLIAMLCNDIYRIGEKLDLGVKYDSSEPSKLELPMPTPIPAEIHYTLLM	3420
.....S.....	
DPMNVFYPGEYVGYLVDAEGGDIYGSYQPTYTYAIIVQEVEREDADNSSFLGKIYQIDIG	3480
.....T.....	
YSEYKIVSSLDLYKFSRPEESSQSRDSAPSTPTSPTEFLTPLGLRSIPPLFSGRESHKT-S	3540
.....D...N...T.....K...SP.	
SKHQSPKKLVNSLPEILKEVTSVVEQAWKLPESEKKIIRRLYLKWHDPKNPFNHDIAN	3600
T..H..R....A.....	
<u>EVFKHLQNEINRLEKQAFLDQNADRASRTFSTSASRFQSDKYSFQRFYTSWNQEATSHK</u>	3660
<u>.....</u>	
<u>SERQQONKEKCPPSAGQTYSQRFVPPFTKSVGNPVEARRWLRQARANFSAARNDLHKNA</u>	3720
<u>.....S.....</u>	
NEWVCFKCYLSTKLALIAADYAVRGKSDKDVKPTALAQKIEEYSQOLEGLTNDVHTLEAY	3780
.....	
GVDSLKTRYPDLLPFPQIPNDRFTSEVAMRMECTACIIIKLENFMQQKV	3830
.....I.....	

Figure 5C

APPROVED	O.G. FIG.	
BY	CLASS	SUBCLASS
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Table 1 ESTs identified by sample-sequencing of the ARSACS critical interval

BAC clone	GenBank #	UniGene	Identity*	Tissue Source
235_I_20	AA987300	IIs. 129092	221/230	neuroendocrine lung carcinoids
235_I_20	AA476635*		249/296	total fetus
235_I_20	AI351876*		272/335	melanocyte, fetal heart, pregnant uterus (pool)
235_I_20	W25994	IIs. 163732	447/464	retina
235_I_20	AI377467	IIs. 163732	257/263	total fetus
235_I_20	AA601007		488/491	schwannoma tumor
235_I_20	AA324964		214/234	cerebellum
235_I_20, 206_I_1	AA897178 ^b		238/238	fetal lung, testis, and B-cell
235_I_20, 206_I_1	RI7106 ^b	IIs. 188560	747/784	brain, adipose tissue
235_I_20, 206_I_1	AB018273 ^b	IIs. 159492	4318/4318	multiple tissue types including brain, CNS, and whole embryo

a: Number of homologous nucleotides between the BAC subclone sequence and the GenBank sequence.

b: AA897178, RI7106, and AB018273 are all contained within *spastin*.

*: AA176335 and AI351876 were homologous to non-overlapping portions of the same M13 subclone sequence.

Figure 6

APPROVED	O.G. FIG.	
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Table 2 Primers for PCR amplification of the human *spastin* ORF

Primer set	Forward primer	Reverse primer	Product size (bp)
1 (SEQ ID NO: 21)	CCTCCAGTACTGTGTTATTGTGAG	CAAGAACTTCCTCAGGGCATC	(SEQ ID NO: 22) 603
2 (SEQ ID NO: 23)	GATGCATCTATACAACATCCGCT	GGTGGAAATAGGTTCTTCTC	(SEQ ID NO: 24) 581
3 (SEQ ID NO: 25)	AAAAATGAGAAATCCAAATGTGCT	GCACTAAGGCTAGGTTTGTGAAG	(SEQ ID NO: 26) 592
4 (SEQ ID NO: 27)	GCTCCTCACTTCCTCTTGTG	CGTGAATTGGCTTCATGATAA	(SEQ ID NO: 28) 602
5 (SEQ ID NO: 29)	AGCAATCAGATTCCAGCAAGC	GATGGGAATGTCAGTGATATGG	(SEQ ID NO: 30) 611
6 (SEQ ID NO: 31)	GGGAGAAAGTTGACAAAGTTGGA	CTTTGGTTTCATCACTGGGAAG	(SEQ ID NO: 32) 624
7 (SEQ ID NO: 33)	TCCAAAGCATTGAACACACCT	CAGGTCCCGTAAGACACTCAG	(SEQ ID NO: 34) 631
8 (SEQ ID NO: 35)	CAATGGGTGCTTTGCTGTTAC	CGAAGAACTCCCGAGAACTCA	(SEQ ID NO: 36) 620
9 (SEQ ID NO: 37)	GCTGGCTGCAAAACAGATACTAC	GCAAAACATGGTTTCAGGCTTA	(SEQ ID NO: 38) 604
10 (SEQ ID NO: 39)	CAACAATCCGCTTCCTTCCAT	ATTATTCGTGCGCAAAAGCTGA	(SEQ ID NO: 40) 651
11 (SEQ ID NO: 41)	TTCCGCGAACTTTTGAAACC	ACACAAAAGTGCTGGCCCTTGC	(SEQ ID NO: 42) 625
12 (SEQ ID NO: 43)	GATGCAAGGCGACAGAAATC	ATACAGCACATTTAGAGCTCCAGT	(SEQ ID NO: 44) 626
13 (SEQ ID NO: 45)	GCATCAGACAGAAATGGTCCAG	GCAATTCAACATATGCAGGAG	(SEQ ID NO: 46) 624
14 (SEQ ID NO: 47)	GTGAAATGGCCACTTTTGCACT	TGATATCAGCAGGGGTCAACAT	(SEQ ID NO: 48) 648
15 (SEQ ID NO: 49)	ACCACACGCAAAACAGTAGCA	GCCATGCATTCTTAAGCCAAAG	(SEQ ID NO: 50) 609
16 (SEQ ID NO: 51)	TGACATTTCCAGCTTTTGCTGA	AGCGGCCACTGATGGATTTAT	(SEQ ID NO: 52) 631
17 (SEQ ID NO: 53)	AAATGATTTTGAGGCACCTTTTG	TTCCACCCAGGATGTCATAAA	(SEQ ID NO: 54) 609
18 (SEQ ID NO: 55)	ACAGTAGACTAAAGCAAGCAAAAGC	ATCAAGAGGAGGATCCAGGTT	(SEQ ID NO: 56) 645
19 (SEQ ID NO: 57)	CATCCTGCCCTATTCTTCCAG	TAAAGCGCAAGGTCTCGTACA	(SEQ ID NO: 58) 618
20 (SEQ ID NO: 59)	TGAGGGCAACAATAGATCC	TCTGCTGTGGGAATAGGATT	(SEQ ID NO: 60) 612
21 (SEQ ID NO: 61)	GCAAAAGCCCTAAGAGAAGGATT	TGCTTTGAGAGCTTTCTCTCAG	(SEQ ID NO: 62) 647
22 (SEQ ID NO: 63)	TGAAAAGAGAAAGATGCTGACAATTC	GTAAGTCTGTCCGGCTGAAGG	(SEQ ID NO: 64) 654
23 (SEQ ID NO: 66)	CATCCCATTTCAGTCAGACA	TTCGTGCTACAAACACATTCAAGA	(SEQ ID NO: 66) 638

Figure 7



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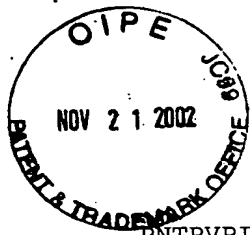
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LOCUS AF193557 11493 bp DNA ROD
DEFINITION Mus musculus saccin gene, complete cds.
ACCESSION AF193557
VERSION AF193557.1 GI:6907043
KEYWORDS .
SOURCE house mouse.
ORGANISM Mus musculus
Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
Mammalia; Eutheria; Rodentia; Sciurognathi; Muridae; Murinae; Mus.
REFERENCE 1 (bases 1 to 11493)
AUTHORS Engert, J.C., Berube, P., Mercier, J., Dore, C., Lepage, P., Ge, B.,
Bouchard, J.P., Mathieu, J., Melancon, S.B., Schalling, M.,
Lander, E.S., Morgan, K., Hudson, T.J. and Richter, A.
TITLE ARSACS, a spastic ataxia common in northeastern Quebec, is caused
by mutations in a new gene encoding an 11.5-kb ORF
JOURNAL Nat. Genet. 24 (2), 120-125 (2000)
MEDLINE 20120709
REFERENCE 2 (bases 1 to 11493)
AUTHORS Engert, J.C., Berube, P., Dore, C., Lepage, P., Ge, B., Hudson, T.J. and
Richter, A.
TITLE Direct Submission
JOURNAL Submitted (08-OCT-1999) Genome Centre, Montreal General Hospital,
1650 Cedar Ave., Montreal, QC H3G 1A4, Canada
FEATURES Location/Qualifiers
source 1..11493
/organism="Mus musculus"
/db_xref="taxon:10090"
mRNA <1..>11493
/product="saccin"
CDS 1..11493
/note="molecular chaperone"
/codon_start=1
/product="saccin"
/protein_id="AAAF31263.1"
/db_xref="GI:6907044"

/translation="MNTFWPGRELVVQWYPFSEDKRHPSLSWLKMVWKNLYIHFSEDL
TLFDEMPLIPRTLLNEDQTCVELIRLRIPSVVILDDTEAQLPEFLADIVQKLGIVL
KRLDTSIQHPLVKKYIHSPLPSAILQIMEKIPLQKLCNQIASLLPTHKDALKRFLASL
TDTSEKEKRIIQELTIFKRINHSSDQGISSYTKLKGCKVLDHTAKLPTDLRLSVSVID
SSDEATIRLANMLKIEKLKTTSCCLKFVLKDIGNAFYTQEEVTQLMLWILENLSSLKNE
NSNVLDWLMLPKFIHMSQGHVVAAGDLDPDIEVLRDLFYNEEEACFPPTIFTSPDIL
HSLRQIGLKNESLKEKDVVQVARKIEALQVSSCQNDVLMKKAKTLLLVLNKNQTLL
QSSEGKMAKKIKWVPACKERPPNYPGSLVWKGDLNLCAPPDMCDAAHAVLVGSSLP
LVESVHVNLEQALSIFTKPTINAVLKHFKTVVDWYTSKTFSEDEYYQFHILLEIYGF
MHDHLSEGKDSFKALKFPWVWTGKNFCPLAQAVIKPTHDLDLQPYLYNVPKTMKAFHQ
LFKACGSIEELTSDHISMVIQVYLKSDQELSEESKQNLHMLNIMRWLYSNQIPAS

Figure 8A



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PNTVPPIYHSRNP SKLVMKPIHECCYCDIKVDDLNDLLED SVEPIILVHEDI PMKTAE
WLKVPCLSTRLINPENMGFEQSGQREPLTVRIKNILEEYPSVSDIFKELLQNADDANA
TECSFMIDMRRNMDIRENLLDPGMAACHGPALWSFNNSEFSDSDFLNITRLGESLKR
EVDKVGKFGFGFNSVYHITDIP IIMSREFMIMFDPNINHISKHIKDRSNPGIKINWSK
QQKRLRKFPNQFKPFIDVFGCQLPLAVEAPYSYNGTLFRLSFRTQQEAKVSEVSSTCY
NTADIYSLVDEFSLCGHRLLIFTQSVNSMYLKYLKIEETNP SLAQDTIIKKKVCPSK
ALNAPVLSVLKEAAKLMKTCSSSNKKLPTDVPKSSCILQITVEEFHHVFRRADLQSP
LFRGPDDDPATLFEMAKSGQSKKPSDEL POKTVDCTTWLICTCMDTGEALKFSLNESG
RRGLGLVPCGAVGVLLHETQE QKWTVKPHIGEVFCYLPLRIKTGLPIHINGCFAVTSNR
KEIWKTDTKGRWNTTFMRHVIVKAYLQALSVLRLDAIGGELTDYTYAVWPD PDLVHD
DFS VICKGFYEDIAHGKGKELTRVFS DSGSMVWSMKNVRFLDD SILQRKDVGSAAFKIF
LKYLKKTGSKNLCAVELPSSVKAGFEEAGCKQILLENTFSEKQFFSEVFFPNIQIEIA
ELRDLPLMNFVLNEKLDEFSGILRVTPCVPCSLEGHPLVLP SRLIHPEGRVAKLFDTKD
GRFPYGSTQDYLNPIILIKLVQLGMAKDDILWDDMLERAESVAEINKSDHAAACLRS
ILLSLIDEKLKIKDPRAKDFAAKYQTI PFLPFLTKPAGFSLEWKGN SFKPETMFAATD
IYTA EYQDIVCLLQPI LNENSHSFRGCGSVSLAVKEFLGLLKKPTVDLVINQLKQVAK
SVDDGITLYQENITNACYKYLHEAVLQNE MAKATIEKLKPF CFI LVENVYVESEKVS
FHLNFEAAPYLYQLPNKYKNFREL FESVGVRSFTVEDFALVLESIDQERGKKQITE
ENFQLCRRIISEGIW SLIREKRQEFCEKNYGKILLPDTNLLLLPAKSLCYND CPWIKV
KDSTVKYCHADIPREVA VKLGAIPKRHKALERYASNICFTALGTEFGQKEKLT SRIKS
ILNAYPSEKEMLKELLQNADDAKATEICFVFDPRQHPVDRI FDDKWAPLQGPALCVYN
NQPFTEDDVRGIQNLGKGTKEGNPCKTGHYGIGFNSVYHITDCPSFISGNDILGIFDP
HARYAPGATSVSPGRMFRDL DADFRTQFSDVLDLYLGNHFKLDNCTMFRFPLRNAEMA
QVSEISSVPSSDRMVQNLLDKLRSDGAELLMFLNHMEKISICEIDKATGGLNVLYSVK
GKITDGDRLKRKQFHASVIDSVTKRQLKDIPVQQITYTMDTEDSEGNLTTWLICNRS
GFSSMEKVS KSVISAHKNQDITLFRGGVAACITHNYKKPHRAFCFLPLSLETGLPFH
VNGHFALDSARRNLWRDDNGVGVRS DWNNSLMTALIAPAYVELLIQLKKRYFPGSDPT
LSVLQNTPIHVVKDTLKKFLSFFPVNRLDLQPDLYCLVKALYSCIHEDMKRLLPVVRA

Figure 8B

APPROVED	O.G. FIG.	
	CLASS	SUBCLASS
BY	DRAFTSMAN	



App No.: 09/693,205
Title: Identification of ARSACS ...
Inventors: Thomas J. Hudson, et al.

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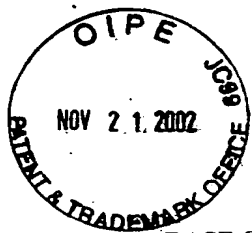
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APPROVED BY DRAFTSMAN	O.G. FIG.	
	CLASS	SUBCLASS

PNIDGSDLHSAVITWINMSTSNKTRPFFDNLLQDELQHLKNADYNITTRKTVAENVY
RLKHLLEIGFNLVYNCDATANLYHCLVDADIPVSYVTPADVRSFLMTFSSPDTNCHI
GKLPCRLOQTNLKLFHSLKLLVDYCFKDAEESEFEVEGLPLLITLDSVLQIFDGKRPK
FLTTHYHELIPSRKDLFMTNTLYLKYSSVLLNCKVAKVFDISSFADLLSSVLPREYKTKN
CAKWKDNFASWLNKAWHFISESVSVTDDQEEPKPAFDVIVDILKDWALLPGTKFTV
STSQLVVPEGDVLIPLSLMHIAVFPNAQSDKVFAHMKAGCIQLALNKICSKDSALVP
LLSCHANIDSPASILKAVHYMVQTSTFRTEKLMENDFEALLMYFNCNLSHLMSQDDI
KILKSLPCYKSIISGRYMSIAKFGTCYVLTKSIPSAEVEKWTQSSSSAFLEEKVHLKEL
YEVLCGPVDDLEVYLKHLPLKIENLSYDAKLEHLIYLNRLASIEEPSEIKEQLFEK
LESLLIHDANNRLKQAKHFYDRTVRVFEVMLPEKLFIPKEFFKKLEQVIKPNQAAF
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DVLQLLWTSCPIPEKATPLSIKEQEGSDAPQEQLQVLNMLNVNLDPPDKVINNC
RNICNITTLDEEMVKTRAKVLRISIYEFLSAEKREFRFQLRGVAFVMVEDGWKLLKPEE
VVINLEYEADFKPYLYKLPLELGTFFHQLFKHLGTEDIISTKQYVEVLSRIFKSSEKQ
LDPNEMRTVKRVVSGLFKSLQNDVSVKVRSDLENARDLALYLPQDGLVKSSILVDDD
APHYKSRIQGNIGVQMLVDLSQCYLGKDHGFHTKLIMLFPQKLRPRLLSSILEEQLE
ETPKVCQFGALCSLQGRQLLLSSEQFITGLIRIMKHENDNAFLANEKAIKRLCKALR
EGLKVSCFEKLQTTLRVKGFNPIPHSRSETFAFLKRFGNAVILLYIQHSDSKDINFL
ALAMTLKSATDNLISDTSYLIAMLCNDIYRISEKLDLGVKYDSSEPSKLELPMPGT
PIPAEIHYTELLMDPMNVFYPGEYVGYLVDAEGGDIYGSYQPTYTYAIIVQEVEREDAD
NTSFLGKIYQIDIGYSEYKIVSSLDLYKFSRPDESSQNRDSAPTTPTSPTEFLTPGLR
SIPPLFSGKESHKSPSTKHHSPRKLKVNALPEILKEVTSVVEQAWKLPESEKKIIRR
LYLKWHDPKNPENHDIANEVFKHLQNEINRLEKQAFLDQADRASRRTFSTSASRFQS
DKYSFQRFYTSWNQEATSHKSERQQQSKEKCPPSAGQTYSQRFVFPPTFKSVGNPVEA
RRWLRQARANFSAARNDLHKNANEWVCFKCYLSTKLALIAADYAVRGKSDKDVKPTAL
AQKIEEYSQOLEGLTNDVHTLEAYGVDSLKTRYPDLLPFPQIPNDRFTSEVAMRVMEC
TACIIIKLENFIQKV"

Figure 8C



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Title: Identification of ARSACS
Inventors: Thomas J. Hudson, et al.

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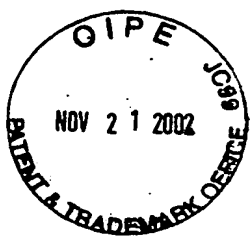
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BASE COUNT 3599 a 2281 c 2387 g 3226 t
ORIGIN

APPROVED	O.G. FIG.		SUBCLASS
	CLASS		
BY			
DRAFTSMAN			

1	atgaatacat	tctggcctgg	tgcagagttg	gtgggttcagt	ggatccatt	tagtgaagac
61	aaacgtcacc	catccctttc	atggccttaag	atgggtttgga	agaatctcta	tatacatttc
121	tcggaagatt	tgacttttatt	tgatgagatg	ccacttatcc	ctagaactct	actgaatgag
181	gaccagacgt	gtgtggaact	catcagactc	aggatcccat	cagtagtcat	tttagatgat
241	gaaactgaag	ctcagcttcc	agaattctta	gcagatattg	tacaaaaact	tggagggatt
301	gtcctgaaaa	gactagatac	ctctattcag	catccacttg	ttaaaaaata	cattcattcc
361	ccactccoga	gtgctatttt	gcagataatg	gagaagatac	ctctacagaa	gttgtgtaat
421	caaatagcat	cattacttcc	aaccacaaaa	gatgctctaa	ggaagttttt	ggccagctta
481	actgatacca	gtgaaaaaga	gaaaagaata	attcaagaat	tgacaatatt	caaaaagaatt
541	aatcactcat	cagatcaagg	gatttctctc	tacacaaaaa	taaaaggatg	taaagttttg
601	gatcataccg	caaagcttcc	aacagatcta	cggctatcag	tttcagtaat	agatagtagt
661	gatgaagcca	ccattcggtt	ggcaaacatg	ttgaaaattg	aaaaattgaa	gactacaagc
721	tgtttaaaagt	ttgttttaaa	agatattgga	aatgcatttt	atacacagga	agaggttaaca
781	caacttatgc	tttggtacct	tgagaatcta	tctctcttta	aaaatgagaa	ttcaaatgtg
841	cttgattggg	taatgccact	aaaattcatt	catatgtccc	agggacatgt	ggtagcagct
901	ggatgatctc	ttgatcctga	tatagaagta	ctaagggatc	tcttttataa	tgaagaagaa
961	gcttgtttcc	cacctacaat	ttttacctca	ccagatatcc	ttcactcttt	gagacagatt
1021	ggcttaaaaa	atgaatccag	tctaaaagaa	aaagatgttg	tacaagtggc	aagaaaaatt
1081	gaagctttac	aggtcagttc	ctgtcagaat	caggatgttc	tcatgaagaa	agccaaaaca
1141	ctcttactgg	tcttgaataa	aaaccagaca	ctcttgacgt	cttctgaagg	gaagatggca
1201	ttgaagaaaa	tcaaattgggt	tccagcctgc	aaggaaaagac	ctccaaatta	tcccgggttc
1261	ttagtctgga	aaggggatct	ctgtaatctt	tgtgcacctc	cagatatgtg	tgatgcggca
1321	catgcagttc	tagtaggctc	ctcacttctc	cttgttgaaa	gtgtccatgt	gaacctggag
1381	caggcgtcca	gcatcttcac	aaagcctact	atcaatgctg	tcttaaaaca	ctttaaaact
1441	gttgttgact	ggtatacttc	aaaaaccttt	agtgatgaag	attactatca	gttccaacat
1501	attttgcttg	aaatttatgg	gttcatgcat	gatcatctga	gtgaagggaa	ggattctttt
1561	aaagccttga	agtttccatg	ggtttggaat	ggcaaaaaact	tttgtcctct	tgcccaggct
1621	gtgataaaagc	caaccatgta	tctggatctt	cagccttatt	tataaatgt	gcctaaaacc
1681	atggcaaaaat	tccaccagtg	gttcaagggt	tgtggctcaa	tagaagagtt	gacatcagat
1741	catatttcca	tggtcattca	gaaagtttat	ctcaaaaagt	accaggagtt	gagtgaagaa
1801	gaaagtaaac	aaaatcttca	tctcatgttg	aatattatga	gatggctcta	tagcaatcag
1861	attccagcaa	gccctaatac	accagttcct	atztatcaca	gcagaaatcc	ttccaaactt
1921	gtcatgaagc	caattcatga	atgctgttat	tgtgacatca	aagttgatga	cctcaatgac
1981	ttgcttgaag	attcagtggg	accaattatc	ttggtacatg	aagatatacc	catgaaaact
2041	gacaagggtg	tgaaagtctc	gtgccttagt	acaagactga	tcaactctga	aaacatgggg
2101	tttgagcagt	cagggcaaaag	agagcctctt	actgtaagga	ttaaaaatat	tttggaagaa
2161	tacccttccg	tgtcagatat	ttttaaagag	ctacttcaaa	atgctgatga	tgcaaatgcc
2221	acagaatgca	gcttcatgat	tgatatgaga	aggaatatgg	acatacggga	aatctcctg
2281	gaccagggga	tggcagcttg	tcatggacct	gctctgtggt	cattcaacaa	ttctgaattc
2341	tcagatttcag	atttcttaaa	cataacgagg	ttaggagagt	ctttaaaaag	gggagaagtt
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2461	atcattatga	gcagagaatt	tatgataatg	tttgatccaa	acataaatca	tatcagcaaa
2521	cacattaaag	atagatcgaa	tcctggaatc	aaaattaatt	ggagtaagca	gcagaaaaga
2581	cttaggaagt	tccccaacca	gttcaaacca	tttatagatg	tatttggtctg	tcagttacct
2641	ttggctgttg	aagctcctta	cagctacaat	ggaactcttt	tccgactgtc	ctttagaaca
2701	cagcaggaag	caaaagttag	tgaagtttag	agtacttgct	acaatactgc	ggatattttac
2761	tccctagtgg	atgaatttag	tctttgtggg	cacagactta	tcattttttac	tcagagtgtg
2821	aactcgatgt	atttgaaata	cttgaaaatt	gaagaaacca	atcctagctt	agcacaagat
2881	acaatcataa	ttaagaaaaa	agtttgcccc	tccaaagcat	tgaatgcacc	agttttaagt
2941	gttttaaaag	aagctgctaa	actcatgaag	acttgtagca	gcagcaacaa	gaagcttccc
3001	acggatgtgc	caaagtcatt	ttgcatctct	cagatcacag	tcgaagaatt	ccaccatgtg
3061	tttaggagga	ttgctgactt	acagtcacca	ctatttcgag	gtccagatga	gtcccagctc
3121	actctctttg	aaatggctaa	atctggccaa	tcaaaaaagc	catcagatga	gttgccacaa
3181	aagacagtag	attgtaccac	atggcttata	tgcacatgca	tggatacagg	agaagctctc
3241	aagttttcct	tgaatgaaag	tgaagaaga	ttagggctgg	ttccttgtgg	ggcagtaggg

Figure 8D



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Inventors: Thomas J. Hudson, et al.

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O.G. FIG.	CLASS	SUBCLASS
	APPROVED BY DRAFTSMAN	

3301	gttctcttgc	atgaaaccca	ggaacagaag	tggaccgtga	aaccacacat	aggagaagtg
3361	ttttgctatt	tacctctacg	aatcaaaaca	gggttgccaa	ttcacatcaa	tgggtgcttt
3421	gctgttactt	caaataaggaa	agaaatctgg	aagacagata	caaaaggctg	atggaatacc
3481	acattcatga	ggcatgtcat	tgtgaaagct	tacttacaag	ccctcagtg	cttacgggac
3541	ctagccattg	gtggtgagct	gactgattat	acttactatg	cagtgtggcc	tgatcctgat
3601	ctagttcatg	atgacttctc	tgtgatctgt	aaaggatttt	atgaagacat	tgctcatggg
3661	aaggggaagg	agttgaccag	agtcttctct	gatgggtcta	tgtgggtttc	catgaagaat
3721	gtgaggtttc	tggatgactc	tatacttcaa	aggaaagatg	ttgggttcagc	agccttcaag
3781	atatttctga	agtacctcaa	gaaaacagga	tccaaaaacc	tctgtgctgt	tgagcttcct
3841	tcttcagtaa	aagcaggatt	tgaagaggct	ggctgtaaac	agatactgct	ggaaaataca
3901	ttttcagaga	aacagttctt	ttcagaagtc	ttctttccta	atatccagga	aattgaagca
3961	gaacttagag	atcctctgat	gaattttgtc	ctaaatgaaa	aacttgatga	gttctcagga
4021	attcttcgtg	ttacctcttg	gtttccttgc	tccttggagg	gccatccttt	ggttttgcct
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4201	ttaggcattg	caaaagatga	tattttgtgg	gatgacatgc	tagagcgtgc	agagtctgta
4261	gctgagatta	ataaaagtga	ccatgctgct	gctgtcttaa	ggagtagtat	tctgctaagc
4321	cttattgatg	agaagctaaa	aataaaggat	cctagagcaa	aggattttgc	tgcaaaatat
4381	caaacaattc	ccttcctccc	atttctaaca	aagccagcag	gtttttcttt	agaatggaaa
4441	gggaacagct	ttaagcctga	aacctggttt	gcagcaactg	acatttacac	agctgaatat
4501	caagatatag	tctgtctttt	gcaaccaatt	cttaatgaaa	attcccattc	ctttagaggc
4561	tgtggttcag	tgtctttggc	tgtaaggag	tttttgggtt	tactaaagaa	gccaacagtt
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4681	taccaggaaa	atatcaccaa	cgcttgctac	aaataacctc	atgaagcagt	attgcagaat
4741	gaaatggcca	aggcaacaat	tattgagaag	ctaaagccat	tttgtttcat	tctagttgag
4801	aatgtatatg	ttgagtcaga	aaaggtttct	tttcaactga	actttgaagc	agcaccatac
4861	ctttatcagt	tacctaacaa	gtataaaaaat	aattttccgtg	agctttttga	aagtgtgggt
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4981	agaggaaaaa	aacaaataac	agaagagaa	tttcagcttt	gccgacgaat	aatcagtgaa
5041	ggcatctgga	gtctcattga	agaaaagaga	caagaatttt	gtgagaaaaa	ttatggcaaa
5101	atattactgc	cagacactaa	cctgctgctg	ctccctgcta	agtcattatg	ctacaatgac
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5341	agaattaaga	gcattctcaa	tgccatctct	tcagaaaagg	aaatgctgaa	agagcttctt
5401	caaaatgctg	atgatgcaaa	ggccacagag	atctgctttg	tgtttgatcc	tagacagcat
5461	cctggttgacc	gaatatttga	tgataagtgg	gccccactgc	aagggccagc	actgtgtgtt
5521	tacaacaacc	agccatttac	agaagatgat	gttagaggaa	ttcagaatct	tgggaaaggc
5581	accaaagaag	ggaatccttg	caaaacagga	cattatggaa	tcggattcaa	ttccgtttat
5641	catattacag	actgcccttc	ttttatttct	ggcaatgaca	tcctgggtat	ttttgatccc
5701	catgccagat	atgcaccagg	agccacatca	gttagccctg	gacgcattgt	tagagatttg
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5881	tcagaaattt	cttccgttcc	atcatcagac	agaatgggtc	agaatctttt	ggacaagtta
5941	cggctctgatg	gggcagaaact	tctaattgtt	ctcaaccaca	tggagaaaaa	atctatttgt
6001	gaaatagata	aggccacagg	aggtctgaat	gtgctctatt	cagtaaaagg	caagatcact
6061	gattggagacc	gattgaaaag	gaagcaattc	cacgcctctg	taattgacag	tgttactaaa
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6181	tctgaaggaa	atctgaccac	atggctcatc	tgtaatagat	caggattttc	aagtatggaa
6241	aaagtatcca	agagtgtaat	atcagctcac	aagaaccaag	atatcaccct	tttcccacgt
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6361	ctgcctctct	ctttggagac	agggtcgcca	tttcatgtga	atggccactt	tgctctagat
6421	tcagccagaa	gaaacttgtg	gcgtgatgat	aatggggttg	gtgttcgaag	tgcttggaat
6481	aatagttaaa	tgacagcatt	aatagcacct	gcatatgttg	agttactaat	ccagttaaaa
6541	aaacgggtatt	tccttggttc	tgacccaaca	ttatcagttt	tacagaacac	acccattcat
6601	gtcgtaaaagg	acacattaaa	gaagtttctg	tccttctttc	cagttaacag	gctggatctg
6661	cagccggact	tatattgctt	agtaaaagca	ctttacagtt	gcattcatga	agacatgaag

Figure 8E



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Inventors: Thomas J. Hudson, et al.

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APPROVED BY DRAFTSMAN	O. G. FIG.	CLASS	SUBCLASS

6721 cgtcttttgc ctgttgttcg ggctccaaat attgatggct cagatttgca ctctgcagtc
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6841 ctacaggatg aattacagca ccttaaaaaat gcagattata acatcacaaac tcgaaaaaca
6901 gtcgcagaga atgtctacag actgaagcac ctgctcttag aaattgggtt caacttgggt
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7081 aattgccata ttgggaagct gccttgtcgt cttcagcaga ctaacctaaa actttttcac
7141 agtttaaaac ttttagttga ttactgtttt aaagatgctg aagaaagtga gtttgaagtt
7201 gagggactgc ccctactcat tacactggac agtgtcttgc agatttttga tggtaaacga
7261 cccaagtttc taacaacata ccatgaatta attccatcgc gtaaagactt gtttatgaac
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Figure 8F



App No.: 09/693,205
Title: Identification of ARSACS ...
Inventors: Thomas J. Hudson, *et al.*

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APPROVED BY DRAFTSMAN	O.G. FIG.	CLASS	SUBCLASS

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11461 aaacttgaaa attttatata acagaaggtg tga

//

Figure 8G



App No.: 09/693,205
Title: Identification of ARSACS ...
Inventors: Thomas J. Hudson, *et al.*

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LOCUS AF193556 12793 bp DNA PRI 07-FEB-2000

DEFINITION Homo sapiens saccin (SACS) gene, complete cds.

ACCESSION AF193556 VERSION AF193556.1 GI:6907041KEYWORDS

SOURCE human.

ORGANISM Homo sapiens Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo.

REFERENCE 1 (bases 1 to 12793) AUTHORS Engert,J.C., Berube,P., Mercier,J., Dore,C., Lepage,P., Ge,B., Bouchard,J.P., Mathieu,J., Melancon,S.B., Schalling,M., Lander,E.S., Morgan,K., Hudson,T.J. and Richter,A. TITLE ARSACS, a spastic ataxia common in northeastern Quebec, is caused by mutations in a new gene encoding an 11.5-kb ORF JOURNAL Nat. Genet. 24 (2), 120-125 (2000) MEDLINE 20120709

REFERENCE 2 (bases 1 to 12793) AUTHORS Engert,J.C., Berube,P., Dore,C., Lepage,P., Ge,B., Hudson,T.J. and Richter,A. TITLE Direct Submission JOURNAL Submitted (08-OCT-1999) Genome Centre, Montreal General Hospital, 1650 Cedar Ave., Montreal, QC H3G 1A4, CanadaFEATURES Location/Qualifiers source 1..12793 /organism="Homo sapiens" /db_xref="taxon:9606" /chromosome="13" /map="between D13S232 and D13S292" mRNA 1..12793 /gene="SACS" /product="saccin" gene 1..12793 /gene="SACS" CDS 77..11566 /gene="SACS" /note="molecular chaperone" /codon_start=1 /product="saccin" /protein_id="AAF31262.1" /db_xref="GI:6907042" /translation="

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KKLDASIQHPLIKYIHSPLPSAVLQIMEKMPLQKLCNQITSLLPTHKDALRKFLASL
TDSSEKEKRIIQELAIKFRINHSSDQGISSYTKLKGCKVLHHTAKLPADLRLSISVID
SSDEATIRLANMLKIEQLKTTSCCLKVLKDIENAFYSHEEVTQLMLWVLENLSSLKNE
NPNVLEWLTPLKFIQISQEQMVSAGELFDPDIEVLKDLFCNEEGTYFPPSVFTSPDIL
HSLRQIGLKNEASLKEKDVVQVAKKIEALQVGACPDQDVLLKAKTLLLVNKNHTLL
QSSEGKMTLKKIKWVPACKERPPNYPGSLVWKGDLNLCAPPDMCDVGHAILIGSSLP
LVESIHVNLEKALGIFTKPSLSAVLKHFKIVVDWYSSKTFSDYDYQFHILLEIYGF
MHDHLNEGKDSFRALKFPWVWTGKKFCPLAQAVIKPIHDLDLQPYLHNVPKTMKFHQ
LFKVCGSIEELTSDHISMVIQKIYKSDQDLSEQESKQNLHMLNIIRWLYSNQIPAS
PNTVPPIHHSKNPSKLIMKPIHECCYCDIKVDDLNDLLEDSEPIILVHEDIPMKTAE
WLKVPCLSTRLINPENMGFEQSGQREPLTVRIKNILEEYPSVSDIFKELLQNADDANA
TECSFLIDMRRNMDIRENLLDPGMAACHGPALWSFNNSQFSDSDFVNITRLGESLKR
EVDKVGKFGLGFNVSYYHITDIPIIMSREFMIMFDPNINHISKHIKDKSNPGIKINWSK
QQKRLRKFPNQFKPFIDVFGCQLPLTVEAPYSYNGTLFRLSFRTQOEAKVSEVSSTCY
NTADIYSLVDEFSLCGHRLIIFTQSVKSMYLYLKIEETNPSLAQDTVIKKKSCSSK
ALNTPVLSVLKEAAKLMKTCSSSNKKLPSDEPKSSCILQITVEEFHHVFRRIADLQSP
LFRGPDDDDPAALFEMAKSGQSKKPSDELSQKTVECTTWLLCTCMDTGEALKFSLSESG
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DFSVICQGFYEDIAHGKKGELTKVSDGSTWVSMKNVRLDSDILKRRDVGSAAFKIF
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FHLNFEAAPYLYQLPNKYKNNFRELFTVGVRSCTVEDFALVLESIDQERGTKQITE
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Figure 9A



App No.: 09/693,205
Title: Identification of ARSACS ...
Inventors: Thomas J. Hudson, et al.

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APPROVED BY DRAFTSMAN	O.G. FIG.	SUBCLASS
	CLASS	

KD TTVKYCHADIPREVAVKLGAVPKRHKALERYASNVCFTTLGTEFGQKEKLTSR IKS
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NQPFTEDDVRGIQNLGKGTKEGNPKYKTGQYGIGFNSVYHITDCPSFISGNDILCIFDP
HARYAPGATSISPGRMFRDLADFR TQFSDVLDLYLGTHFKLDNCTMFRFPLRNAEMA
KVSEISSVPASDRMVQNLLDKLRSDGAELLMFLNHMEKISICEIDKSTGALNVLYSVK
GKITDGDRLKRKQFHASVIDSVTKKRQLKDIPVQQITYTMDTEDSEGNLT TWLICNRS
GFSSMEKVS KSVISAHKNQDITLFFPRGGVAACITHNYKKPHRAFCFLPLSLETGLPFH
VNGHFALDSARRNLWRDDNGVGVRSDWNNSLMTALIAPAYVELLIQLKKRYFP GSDPT
LSVLQNTPIHVVKDTLKKFLSFFPVNRLDLQPDLYCLVKALYNCIHEDMKRLLPVVRA
PNIDGSDLHSAV IITWINMSTSNKTRPFFDNLLQDELQHLKNADYNITTRKTVAENVY
RLKHLLLEIGFNLVYNCD ETANLYHCLIDADIPVS YVTPADIRSFLMTFSSPD TNCHI
GKLPCRLQQTNLKLFHSLKLLVDYCFKDAEENEIEVEGLPLLITLDSVLQTFDAKRPK
FLT TYHELIPSRKDLFMNTLYLKYSNILLNCKVAKVFDISSFADLLSSVLPREYKTKS
CTKWKDNFAS ESWLKNAWHFISESVSVKEDQEETKPTFDIVVDTLKD WALLPGTKFTV
SANQLVVP EGDVLLPLSLMHIAVFPNAQSDKVFHALMKAGCIQLALNKICSKDSAFVP
LLSCHTANIESPTSILKALHYMVQTSTFRAEKL VENDORFEALLMYFNCNLNHLMSQDDI
KILKSLPCYKSI SGRYVSIGKFGTCYVLT KSI PSAEVEKWTQSSSAFLEEKIHLKEL
YEVIGCVPVDDLEVLYLKHLLPKIENLSYDAKLEHLIYLKNRLSSAEELSEIKEQLFEK
LESLLI IHDANSRLKQAKHFYDRTVRVFEV MLPEKLFIPNDFFKKLEQLIKPKNHVTF
MTSWVEFLRNIGLKYILSQQQLLQFAKEISVRANTENWSKETLQNTVDILLHHIFQER
MDLLSGNFLKELSLIPFLC PERAPAEFIRFHPQYQEVNGTLPLIKFNGAQVNPKFKQC
DVLQLLWTSCPI LPEKATPLS I KEQEGSDLG PQEQLEQVLNMLNVNLD PPLDKVINNC
RNICNITTLDEEMVKTRAKVLRSIYEFLSAEKREFR FQLRGVAFVMVEDGWKLLKPEE
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RWLRQARANFSAARNDLHKNA NEWVCFKCYLSTKLALIAADYAVRGKSDKDVKPTALA
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BASE COUNT 4163 a 2256 c 2487 g 3887 tORIGIN
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Figure 9B



App No.: 09/693,205
Title: Identification of ARSACS . . .
Inventors: Thomas J. Hudson, et al.

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APPROVED BY	O.G. FIG.	
	CLASS	SUBCLASS
DRAFTSMAN		

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Figure 9C



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Inventors: Thomas J. Hudson, et al.

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APPROVED	O.G. FIG.	
	BY	DRAFTSMAN
	CLASS	SUBCLASS

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gaaaaattat	ggcaagatat	tattgccaga	tactaatctt	atgcttctcc	ctgctaaatc
gttatgctac	aatgattgcc	cttggataaa	agtaaaggat	accactgtaa	aatattgtca
tgctgacata	cccagggaa	tagcagtaaa	actaggagca	gtcccaaagc	gacacaaagc
cttagaaaga	tatgcatcca	atgtctgttt	tacaacactt	ggcacagaat	ttgggcagaa
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cttttcaagt	atggagaaa	tatctaaaa	tgatcatatca	gtcacaaga	accaagatat
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attttttgac	aattttactac	aggatgaatt	acaacacctt	aaaaatgcag	attataatat
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Figure 9D



App No.: 09/693,205
Title: Identification of ARSACS ...
Inventors: Thomas J. Hudson, et al.

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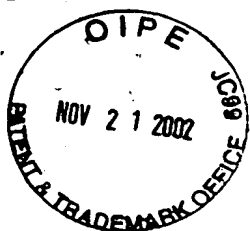
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APPROVED BY DRAFTSMAN	O.G. FIG.
	CLASS SUBCLASS

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atataagacc	aaaagttgca	caaagtggaa	agacaatttt	gcaagtgaat	cttggcttaa
gaatgcatgg	cattttatta	gtgaatctgt	aagtgtgaaa	gaagatcagg	aagaaacaaa
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tcctttgttg	tcattgtcac	cagcaaatat	agagagcccc	acaagcatct	tgaaggctct
acattatatg	gtccaaactt	caacatttag	agcagaaaaa	ttagtagaaa	atgattttga
ggcacttttg	atgtatttca	actgcaattt	gaatcatttg	atgtcccaag	atgatataaa
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acaatcatca	tcattctgcat	ttcttgaaga	aaaaatacac	ttaaaagaac	tatatgaggt
gattgggtgt	gtacctgtag	atgatcttga	ggtatatttg	aaacacctct	taccaaaaat
tgaaaatctc	tcttatgatg	caaaattaga	gcacttgatc	taccttaaga	atagattatc
aagtgctgag	gaattatcag	agattaagga	acaacttttt	gaaaaactgg	aaagtttatt
gataatccat	gatgctaaca	gtagactaaa	gcaagcaaag	catttctatg	atagaactgt
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tgcgctttac	ctcccaagcc	aggatggtag	attggtaaag	tcaagcatct	tagtggttga
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tctcagccag	tgctacttag	ggaaagacca	tggatttcac	actaagttga	taatgctctt
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tgcttttctg	gccaatgaag	aaaaagccat	aagactttgc	aaagccctaa	gagaaggatt
gaaagtatcc	tgctttgaaa	agcttcaaac	aacattaaga	gttaaagggt	ttaatcctat
tccccacagc	agaagtgaag	cttttgcttt	tttgaagcga	tttggtaatg	cagtcactct
gctctacatt	caacattcag	acagtaaaga	cattaatttc	ctgttagcac	tggcaatgac
tcttaaatca	gcaactgaca	atgtgatttc	tgacacttca	tatttaattg	ctatgctagg
atgcaatgat	atttacagga	ttggtgagaa	acttgacagt	ttaggagtga	aatatgactc
ttcggagcca	tcaaaactgg	aacttccaat	gcctggcaca	ccaattcctg	ctgaaattca
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tcagatagat	attggttata	gtgaatataa	aatagttagc	tctcttgatc	tgtataagtt
ttcaagacct	gaggaaagct	ctcaaagcag	ggacagtgtc	ccttctacac	caaccagccc

Figure 9E



App No.:
Title:
Inventors:

09/693,205
Identification of ARSACS ...
Thomas J. Hudson, *et al.*

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APPROVED BY DRAFTSMAN	O.G. FIG.	SUBCLASS
	CLASS	

cactgagttc ctcacccctg gcctgagaag cattcctcct cttttctctg gtagagagag
ccacaagact tctttccaaac atcagtcctc caaaaagctt aagggttaatt ctttaccaga
aatcttataaa gaagtgaacat ctgtggtgga gcaagcatgg aagcttccag aatcggaacg
aaaaaagatt attaggcggg tgtatttgaa atggcatcct gacaaaaatc cagagaacca
tgacattgcc aatgaagttt ttaaaccatt gcagaatgaa atcaacagat tagaaaaaca
ggctttttcta gatcaaaatg cagacagggc ctccagacga acattttcaa cctcagcatc
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acagacttac tctcaaagggt tctttgttcc tcccactttc aagtcggttg gcaatccagt
ggaagcacgc agatggctaa gacaagccag agcaaaactc tcagctgcca ggaatgacct
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Figure 9F